DISORDERS COVERED BY THE PROGRAM

Effective January 1, 2006, Utah newborns are screened for the following disorders:

METABOLIC DISORDERS:

- Biotinidase deficiency: a recessive disorder of biotin metabolism
- Galactosemia: a recessively inherited genetic disorder in which the individual is completely or partially incapable of normal metabolism of galactose due to a deficiency of the galactose-1-phosphate uridyltransferase enzyme

Fatty Acid Oxidation Disorders: a recessive disorder resulting from an enzyme deficiency needed for the break down of fatty acids

- Carnitine uptake/transport defects
- Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Carnitine-Acylcarnitine Translocase Deficience
- Carnitine Palmitoyl Transferase-1 Deficiency

Amino acid disorders: a recessive disorder resulting from an enzyme deficiency needed for amino acid metabolism or transport

- Arginase Deficiency
- Argininosuccinate lyase deficiency (ASA)
- Citrullinemia
- Homocystinuria
- Hyperphenylalanemia, including phenylketonuria
- Tyrosinemia

Organic Acid Disorders: a recessive disorder resulting from an enzyme deficiency in the intermediary metabolism of amino acids or fatty acids

- Beta-ketothiolase deficiency
- Glutaric acidemia, Type 1
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric acidemia
- Malonic aciduria
- Maple syrup urine disease
- Methylmalonic acidemias
- Propionic acidemia
- 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency
- 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
- 2-Methylbutyryl CoA dehydrogenase deficiency
- Multiple carboxylase deficiency

ENDOCRINE DISORDERS:

- Congenital adrenal hyperplasia (CAH): a genetic disorder in which there are defects in the enzymes of the adrenal cortex required for the biosynthesis of adrenal corticosteroids
- Congenital hypothyroidism: a disorder in which the newborn is unable to secrete or produce thyroxine normally

HEMOGLOBIN DISORDERS:

- Sickle cell disease and other hemoglobinopathies: a recessively inherited genetic defect of the structure of hemoglobin found in red blood cells

These are disorders which may have significant mortality and morbidity when not diagnosed presymptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders.

Newborn
Screening Program

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